



## PNPLA6 gene

patatin like phospholipase domain containing 6

### Normal Function

The *PNPLA6* gene provides instructions for making a protein called neuropathy target esterase (NTE). The NTE protein is involved in the breakdown (metabolism) of certain fats (lipids). Specifically, NTE breaks down a fat called lysophosphatidylcholine, which is one of several compounds found in the outer membranes surrounding cells. The correct levels of these compounds are critical to the stability of the cell membranes. NTE helps regulate the levels of lysophosphatidylcholine, which can be damaging to cells in high amounts.

The NTE protein is found most abundantly in the nervous system. It plays an important role in maintaining the stability of the membranes surrounding nerve cells (neurons) and of these cells' specialized extensions, called axons, that transmit nerve impulses. NTE may also play a role in the release of hormones from the pituitary gland, a process that requires particular changes in the cell membrane and appears to involve the lipids found there. The pituitary gland is located at the base of the brain and produces several hormones, including those that help direct sexual development and growth.

### Health Conditions Related to Genetic Changes

#### Boucher-Neuhäuser syndrome

More than a dozen mutations in the *PNPLA6* gene have been found to cause Boucher-Neuhäuser syndrome, a disorder characterized by coordination and balance problems (ataxia), vision impairment, and delayed puberty. The mutations are thought to impair the function of the NTE protein. Researchers are unsure how such a reduction in function leads to the signs and symptoms of the condition. They speculate that impairment of lysophosphatidylcholine metabolism alters the balance of compounds in the cell membrane. This imbalance may damage axons, leading to the movement and vision problems that characterize Boucher-Neuhäuser syndrome. The imbalance is also thought to impair the release of hormones involved in sexual development, accounting for the delayed puberty in affected individuals.

#### other disorders

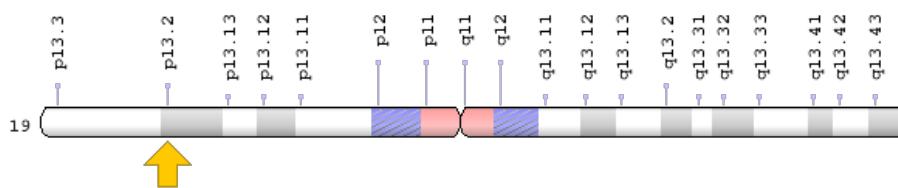
Mutations in the *PNPLA6* gene cause a continuous spectrum of neurological conditions called *PNPLA6*-related disorders. Conditions in this group include Boucher-Neuhäuser syndrome (described above), Gordon Holmes syndrome, Oliver-McFarlane syndrome, Laurence-Moon syndrome, and spastic paraplegia type 39.

*PNPLA6*-related disorders feature combinations of overlapping signs and symptoms, including ataxia, muscle stiffness (spasticity), abnormally fast (brisk) reflexes, reduced sensation in the extremities (peripheral neuropathy), cognitive impairment, eye abnormalities, impaired vision, hair abnormalities, reduced production of the hormones that direct sexual development (hypogonadotropic hypogonadism), and reduced function of the pituitary gland (hypopituitarism). It is unknown how mutations in a single gene cause such a wide range of disorders.

### Chromosomal Location

Cytogenetic Location: 19p13.2, which is the short (p) arm of chromosome 19 at position 13.2

Molecular Location: base pairs 7,534,152 to 7,561,767 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

### Other Names for This Gene

- BNHS
- iPLA2delta
- LNMS
- NTE
- NTEMND
- OMCS
- patatin-like phospholipase domain-containing protein 6
- SPG39
- SWS

## **Additional Information & Resources**

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): The Lipid Bilayer  
<https://www.ncbi.nlm.nih.gov/books/NBK26871/>

### GeneReviews

- PNPLA6-Related Disorders  
<https://www.ncbi.nlm.nih.gov/books/NBK247161>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PNPLA6%5BTIAB%5D%29+OR+%28patatin+like+phospholipase+domain+containing+6%5BTIAB%5D%29%29+OR+%28%28NTE%5BTIAB%5D%29+OR+%28patatin-like+phospholipase+domain-containing+protein+6%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+NOT+%28non-targeted+effects%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

### OMIM

- PATATIN-LIKE PHOSPHOLIPASE DOMAIN-CONTAINING PROTEIN 6  
<http://omim.org/entry/603197>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_PNPLA6.html](http://atlasgeneticsoncology.org/Genes/GC_PNPLA6.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=PNPLA6%5Bgene%5D>
- HGNC Gene Family: Patatin like phospholipase domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/466>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=16268](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=16268)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/10908>
- UniProt  
<http://www.uniprot.org/uniprot/Q8IY17>

## Sources for This Summary

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*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4245359/>
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